Gene Set BenchMark

Bahman Afsari[1] and Elana J. Fertig[1]

[1]The Sidney Kimmel Comprehensive Cancer Center,
Johns Hopkins University School of Medicine

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1 Introduction

The `GSBenchMark` contains eleven expression datasets representative of different diseases. The package also contains a list of pathways and their associated gene targets. Together with these datasets and the pathways provide a benchmark for machine learning and pathway analyses, most of them used previously in [1].

2 Datasets

Benchmark datasets and pathway targets were downloaded from supplemental files and sources cited in [1]. These datasets covers different diseases: Leukaemia [2], Marfan [3], Melanoma [4], Prostate [5], Sarcoma [6], Head and neck cancer [7], response to breast cancer treatmetn [8], Bipolar disorder [9]. We also added datasets for two new diseases: Parkinson's disease [10], and Melanoma cancer[4]. We did not include two of the datasets mentioned in [1]: First, the famous Leaukemia data set in [11] which is available through package `golubEsets`. Secondly, the data presented in paper [12] because the data were not available to us. These data were converted from Matlab to R for this package.

First, we load the library:

```
> require(GSBenchMark)
```

2.1 Pathway Data

`GSBenchMark` contains a list of the pathways.

```
> data(diracpathways)
> class(diracpathways)

[1] "list"

> names(diracpathways)[1:5]

[1] "DEATHPATHWAY"       "TCAPOPTOSISPATHWAY" "CCR3PATHWAY"
[4] "NEUTROPHILPATHWAY"  "ALTERNATIVEPATHWAY"

> class(diracpathways[[1]])

[1] "character"

> diracpathways[[1]]

      "BID"     "TRAF2"  "TNFRSF25"    "NFKBIA"     "NFKB1"   "TNFSF12"     "CASP6"

    "CASP3"     "CASP9"     "CASP7"      "BCL2"     "CASP8"      "CHUK"     "CFLAR"

     "DFFA"      "DFFB"      "RELA"      "CYCS"      "LMNA"      "GAS2"      "FADD"

    "BIRC4"     "BIRC3"     "BIRC2"     "TRADD" "TNFRSF10A"    "CASP10"   "TNFSF10"

"TNFRSF10B"     "RIPK1"     "APAF1"   "MAP3K14"    "SPTAN1"

> pathways = diracpathways;
```

The variable `diracpathways` contains the pathways genes. It is a list. Each element represents a pathway with its name. Each elements contains a list of characters which represent the genes in the pathway.

2.2 Gene Expression Datasets

Now, we load the datasets names:

```
> data(GSBenchMarkDatasets)
> print(GSBenchMark.Dataset.names)
```

```
 [1] "leukemia_GSEA"          "marfan_GDS2960"         "melanoma_GDS2735"
 [4] "parkinsons_GDS2519"     "prostate_GDS2545_m_nf" "prostate_GDS2545_m_p"
 [7] "prostate_GDS2545_p_nf" "sarcoma_data"           "squamous_GDS2520"
[10] "breast_GDS807"          "bipolar_GDS2190"
```

Here is a summary of the datasets:

```
> for(i in 1: length(GSBenchMark.Dataset.names))
 {
   DataSetStudy = GSBenchMark.Dataset.names[[i]]
   data(list=DataSetStudy)
   cat("Data Set",DataSetStudy,"\n")
   print(phenotypesLevels)
   print(table(phenotypes))
 }
```

```
Data Set leukemia_GSEA
    0     1
"AML" "ALL"
phenotypes
 0  1
24 24
Data Set marfan_GDS2960
        0         1
"non-MFS"     "MFS"
phenotypes
 0  1
41 60
Data Set melanoma_GDS2735
           0            1
    "Normal" "metastasis"
phenotypes
 0  1
23 23
Data Set parkinsons_GDS2519
            0            1
     "Normal" "Parkinson's"
phenotypes
 0  1
22 50
Data Set prostate_GDS2545_m_nf
           0            1
    "normal" "metastasis"
phenotypes
```

```
 0  1
18 25
Data Set prostate_GDS2545_m_p
          0           1
   "primary" "metastasis"
phenotypes
 0  1
65 25
Data Set prostate_GDS2545_p_nf
        0         1
 "normal" "primary"
phenotypes
 0  1
18 65
Data Set sarcoma_data
     0      1
 "LMS" "GIST"
phenotypes
 0  1
31 37
Data Set squamous_GDS2520
       0        1
"Normal"  "HNSCC"
phenotypes
 0  1
22 22
Data Set breast_GDS807
               0                1
    "Responsive" "Non-responsive"
phenotypes
 0  1
32 28
Data Set bipolar_GDS2190
        0         1
 "Normal" "Bipolar"
phenotypes
 0  1
31 30
```

The data consist of three variables: `exprsdata`, `phenotypes`, and `phenotypesLevels`. `exprsdata` consists of gene expressions. `phenotypes` contains the sample labels: "0" indicates less dangarous and "1" more dangerous phenotype. `phenotypesLevels` makes the connection between "0" and "1" with the real label names e.g. "Normal" and "Parkinson's". `GSBenchMark` requires the rownames of gene expression varaible represent the gene names, *i.e.* they are represented in the pathway information variable.

2.3 Matching pathway targets to gene expression datasets

One can extract the gene names by:

```
> genenames = rownames(exprsdata);
> genenames[1:10]
 [1] "DDR1"   "RFC2"   "HSPA6"  "PAX8"   "GUCA1A" "UBA7"   "THRA"   "PTPN21" "CCL5"
[10] "CYP2E1"
```

Also, it is possible that some of the genes in a pathway are not represented in the expression data. We prune them as the following:

```
> prunedpathways = lapply(pathways, function(x) intersect(x, genenames))
> emptypathways = which(sapply(prunedpathways, length) < 2)
> if (length(emptypathways) > 0) {
        warning(paste("After pruning the pathways, there exist pathways with zero or one
            paste(names(emptypathways), collapse = ","), "\n"))
        diracpathwayPruned= prunedpathways[-emptypathways]
 }else {
        diracpathwayPruned = prunedpathways
 }
> cat("Number of pathways before pruning ",length(pathways),"\n")

Number of pathways before pruning  249

> cat("Number of pathways after pruning ",length(diracpathwayPruned))

Number of pathways after pruning  249
```

`phenotypes` is a factor with with levels ("0","1") where "1" indicates more dangerous phenotype. For real labels, one can look at `phenotypesLevels`

```
> summary(phenotypes)
 0  1
31 30

> phenotypesLevels

        0         1
 "Normal" "Bipolar"
```

3 System Information

Session information:

```
> toLatex(sessionInfo())
```

- R version 4.5.1 Patched (2025-08-23 r88802), x86_64-pc-linux-gnu
- Locale: LC_CTYPE=en_US.UTF-8, LC_NUMERIC=C, LC_TIME=en_GB, LC_COLLATE=C, LC_MONETARY=en_US.UTF-8, LC_MESSAGES=en_US.UTF-8, LC_PAPER=en_US.UTF-8, LC_NAME=C, LC_ADDRESS=C, LC_TELEPHONE=C, LC_MEASUREMENT=en_US.UTF-8, LC_IDENTIFICATION=C
- Time zone: America/New_York
- TZcode source: system (glibc)
- Running under: Ubuntu 24.04.3 LTS
- Matrix products: default
- BLAS: /home/biocbuild/bbs-3.22-bioc/R/lib/libRblas.so
- LAPACK: /usr/lib/x86_64-linux-gnu/lapack/liblapack.so.3.12.0
- Base packages: base, datasets, grDevices, graphics, methods, stats, utils
- Other packages: GSBenchMark 1.30.0
- Loaded via a namespace (and not attached): compiler 4.5.1, tools 4.5.1

4 Literature Cited

References

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